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Current Strategies for the Biochemical Diagnosis and Monitoring of Mitochondrial Disease

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Edited by
Iain Hargreaves

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Mitochondrial disease constitutes a complex and heterogeneous group of disorders resulting from a defect in mitochondrial respiratory chain (MRC) enzyme activity. In view of the dual regulation of the MRC, exercised by both the mitochondrial and nuclear genome, mutations in either mitochondrial or nuclear DNA can result in a MRC deficiency. Whilst a single organ can be affected, MRC disorders often result in a multi-organ system presentation with prominent neurological and myopathic features. The diagnosis of MRC disorders can be complex, and requires a coordinated interplay of a number of disciplines. However, biochemical determination of metabolites in blood, cerebral spinal fluid (CSF) and/or urine are generally considered to be first-line investigations for the diagnosis of these disorders, although they lack sensitivity and specificity. Furthermore, there is a lack of consensus on the overall utility of monitoring other biochemical parameters, which may be of diagnostic value. For example, although oxidative stress may contribute to the pathogenesis of mitochondrial disorders, few centers monitor this as part of their diagnostic repertoire. Therefore, the purpose of this Special Issue was to highlight potential biomarkers of mitochondrial disease and to discuss the appropriateness of biochemical markers to monitor disease progression and therapeutic intervention.

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